

What is claimed is:

1. Isolated nucleic acid encoding a mammalian SCA2 polypeptide.
2. The isolated nucleic acid of Claim 1 which is DNA.
3. The isolated nucleic acid of Claim 2, wherein the DNA is cDNA.
4. The isolated nucleic acid of Claim 2 which encodes at least about 10 contiguous amino acids set forth in SEQ ID NO: 3 or at least about 10 contiguous amino acids set forth in SEQ ID NO:5.
5. The isolated nucleic acid of Claim 2 which hybridizes under high stringency conditions to nucleotides 1 - 516 of SEQ ID NO:1, nucleotides 163-4098 of SEQ ID NO:2 or SEQ ID NO:4.
6. The isolated nucleic acid of Claim 2, which has substantially the same nucleotide sequence as nucleotides 1 - 516 of SEQ ID NO:1, nucleotides 163-4098 of SEQ ID NO:2 or SEQ ID NO:4.
7. A vector comprising the isolated nucleic acid of Claim 2.
8. The isolated nucleic acid of Claim 2 which hybridizes under high stringency conditions to nucleotides 163-4098 of SEQ ID NO:2.
9. The isolated nucleic acid of Claim 2, which has substantially the same nucleotide sequence as nucleotides 163-4098 of SEQ ID NO:2.
10. An isolated oligonucleotide comprising at least 15 nucleotides capable of specifically hybridizing with a sequence of nucleic acids of the nucleotide sequence set forth in SEQ ID NO:2 or the nucleotide sequence set forth in SEQ ID NO:4.

11. The isolated oligonucleotide of Claim 10 which is labeled with a detectable marker.
12. The isolated nucleic acid of Claim 2, wherein the DNA has at least 90% homology to the SCA2 coding portion set forth in SEQ ID NO:2.
13. The isolated nucleic acid of Claim 1, encoding a mouse SCA2 polypeptide.
14. The isolated nucleic acid of Claim 1, which is DNA.
15. The isolated nucleic acid of Claim 14, wherein said DNA is cDNA.
16. The isolated nucleic acid of Claim 14, which hybridizes under high stringency conditions to the SCA2 coding portion of SEQ ID NO:4.
17. The isolated nucleic acid of Claim 14, which has at least 90% homology to the SCA2 coding portion set forth in SEQ ID NO:4.
18. A vector comprising the isolated nucleic acid of Claim 14.
19. An isolated nucleic acid comprising nucleotides 163-657 of SEQ ID NO:2.
20. An isolated nucleic acid comprising nucleotides 724-4098 of SEQ ID NO:2.
21. An isolated nucleic acid comprising at least about 15 contiguous nucleotides from nucleotides 163-657 of SEQ ID NO:2, or the nucleotides complementary thereto.
22. An isolated nucleic acid consisting of at least about 15 continuous nucleotide from nucleotides 724-4098 of SEQ ID NO:2, or the nucleotides complementary thereto.

23. An isolated nucleic acid comprising nucleotides 163-4098 of SEQ ID NO:2.
24. An isolated nucleic acid comprising SEQ ID NO:4.
25. A single strand DNA primer comprising a nucleic acid sequence derived from the isolated nucleic acid of Claim 1.
26. The single strand DNA primer of Claim 25 wherein the nucleic acid comprises the nucleic acid sequence set forth in SEQ ID NO:2 or the nucleic acid sequence set forth in SEQ ID NO:4.
27. A method of detecting the presence or absence of a CAG repeat region in the SCA2 gene on human chromosome 12 at locus q 24.1 in an individual comprising the steps of obtaining a nucleic acid sample from an individual and detecting the presence of the CAG repeat region in the sample.
28. The method of Claim 27 wherein said individual is suspected of having SCA2.
29. The method of Claim 27 wherein the individual has an extended CAG repeat.
30. A method of diagnosing spinocerebellar ataxia type 2 in a human nucleic acid sample comprising the steps of:
amplifying said nucleic acid sample with a first primer and a second primer by
polymerase chain reaction, wherein said first primer hybridizes to a region of
nucleotides 303 to 657 of SEQ ID NO:2 and said second primer hybridizes to a
region of nucleotides 723 to 890 of SEQ ID NO:2;
obtaining an amplification product of said nucleic acid sample by said polymerase chain
reaction; and
measuring a number of CAG repeats in said amplification product,

wherein a normal number of CAG repeats in said nucleic acid sample would be negative for spinocerebellar ataxia type 2.

31. A method of diagnosing spinocerebellar ataxia type 2 in a human nucleic acid sample comprising the steps of:
amplifying said nucleic acid sample with a first primer and a second primer by polymerase chain reaction, wherein said first primer hybridizes to a region of nucleotides 303 to 657 of SEQ ID NO:2 and said second primer hybridizes to a region of nucleotides 723 to 890 of SEQ ID NO:2;
obtaining an amplification product of said nucleic acid sample by said polymerase chain reaction; and
measuring a number of CAG repeats in said amplification product,
wherein a number of 35 or more CAG repeats in said nucleic acid sample would be indicative of said spinocerebellar ataxia type 2 and a number of 15-24 CAG repeats in said nucleic acid sample would be negative for spinocerebellar ataxia type 2.
32. The method of Claim 30, wherein said first primer has a sequence as set forth in nucleotides 637 to 656 of SEQ ID NO:2.
33. The method of Claim 30, wherein said second primer has a sequence complementary to that of nucleotides 764 to 783 of SEQ ID NO:2.
34. The method of Claim 30, wherein said number of CAG repeats is measured by gel electrophoresis.
35. The method of Claim 30, wherein said number of CAG repeats is measured by sequencing said amplification product.

36. The method of Claim 30, wherein said number of CAG repeats is measured by hybridizing a probe to said amplification product, wherein said probe has a sequence comprising greater than 22 CAG repeats.
37. The method of Claim 30, wherein the nucleic acid sample is genomic DNA.
38. The method of Claim 30, wherein the nucleic acid sample is cDNA.
39. A method of diagnosing SCA2 in a human sample comprising identifying the presence of a CAG repeat in a nucleic acid sample from chromosome 12 at locus q 24.1, wherein the presence of a larger number of CAG repeats than exists in a normal population is indicative of SCA2.
40. A method of diagnosing spinocerebellar ataxia type 2 in a human nucleic acid sample comprising the steps of:
amplifying said nucleic acid sample with a first primer and a second primer by polymerase chain reaction, wherein said first primer hybridizes to a region of nucleotides 163 to 657 of SEQ ID NO:2 and said second primer hybridizes to a region of nucleotides 724-4098 of SEQ ID NO:2;
obtaining an amplification product of said nucleic acid sample by said polymerase chain reaction; and
measuring a number of CAG repeats in said amplification product,
wherein thirty-two or more CAG repeats in said nucleic acid sample is indicative of said spinocerebellar ataxia type 2 and a normal number of CAG repeats in said nucleic acid sample would be negative for spinocerebellar ataxia type 2.
41. A method of diagnosing SCA2 in a human sample comprising obtaining a nucleic acid sample from a human and identifying the presence of a CAG repeat in a nucleic acid sample from chromosome 12 at locus q 24.1, wherein the presence of a larger number of CAG repeats than exists in a normal population is indicative of SCA2.